ABSTRACT

Renal agenesis is relatively common, genetically determined, disease. Genes which lead to its origin are still unconfirmed.

Subject of this study was to perform molecular-genetic analyses of two genes, which are candidate for origin of renal agenesis in humans, genes coding tyrosine kinase receptor RET and neurotrophic factor GDNF. Mutation analysis of 20 exons of RET and 3 exons of GDNF in group of 20 patients with diagnosed unilateral renal agenesis has been done. Numeric changes were also investigated. The aim of this work was to identify potential mutation of RET and GDNF genes and contribute to their association with renal agenesis formation.

No pathogenic mutation has been found in the group of patients, only three known single-point polymorphisms in *RET* gene have been detected. Polymorphism rs1800860 (GCG-GCA, Ala-Ala 432) is situated in exon 7 and has been found in 9 of total 20 patients, thereof in two cases in homozygous state. Polymorphism rs1800861 (CTT-CTG, Leu-Leu 769) is located in exon 13 and has been identified in 3 of 20 patients, thereof one patient was homozygote for minority allele G. Polymorphism rs1800863 (TCC-TCG; Ser-Ser) in 15. exon has been found in 5 patients, at any time in heterozygous state. All cases are about common polymorphisms with frequency of minority allele in population higher then 10%.

The results of this study did not confirm increased occurrence of *RET* and *GDNF* mutations in patients with renal agenesis and they couldnôt support association of these two genes with disease origin. However given limited size of group of patients it is impossible to disprove connection between RET/GDNF signalling complex and renal agenesis.

Key words: Renal agenesis, RET, GDNF, polymorphism.